

Investigating Birth Defects

Caleb Cheung, NBCT

Frick Middle School, Oakland, California

In collaboration with James Kucik, National Center on Birth Defects and Developmental Disabilities, Centers for Disease Control and Prevention

Disclaimer: The findings and conclusions in this report are those of the author(s) and do not necessarily represent the views of the Centers for Disease Control and Prevention.

Investigating Birth Defects

Caleb Cheung, NBCT
Frick Middle School
Oakland, CA

Summary

In this lesson, students take on the role of epidemiologists and investigate mystery birth defects occurring around their city or town. Students form groups, then research and present information about various birth defects and developmental disabilities to their peers. In the process, they learn about possible causes, treatments, and prevention methods for these birth-related conditions.

Because of the vocabulary students will encounter on websites while conducting their research, this lesson would fit best after students have studied the human body, human reproduction, cells, and genetics. However, extra time can be allocated for understanding difficult vocabulary if it has not been covered previously.

Learning Outcomes

- Students will learn about the most common birth defects in the United States by researching and presenting the information in groups to their peers.
- Students will create a chart to record information about birth defects.
- Students will apply their knowledge of birth defects to diagnose reported cases of health-related conditions in babies.

Materials

1. Computers with Internet access
2. Overhead or LCD projector

Total Duration

3 hours

Procedures

Teacher Preparation

Before the lesson, you may want to read the websites below to familiarize yourself with general background information about birth defects.

Download and make copies of the “Letter from the Center for Disease Defense and Prevention (CDD),” “Birth Defects and Developmental Disabilities Chart,” and “Case Presentation Rubric” for all the students in the class and enough “Case Files” for each group (one per group). Also, modify the CDD letter to match the date, name, and address of your school. If you do not have access to an LCD projector, make an overhead transparency of the “Birth Defects and Developmental Disabilities Chart” for reviewing the answers with students.

An overhead is used in the description for writing down ideas and responses from students. If one is not available, a white board, chalk board, or computer and LCD projector can be used instead.

Throughout the lesson, the term “health-related condition” is used as a more general and appropriate term instead of the more colloquial “disease” or “illness.”

Web Resources

Title: March of Dimes—Birth Defects

URL: www.marchofdimes.com/pnhec/4439.asp

Description: This is an organization focused on birth defects prevention. The descriptions of birth defects, developmental disabilities, and other disorders on this website are the least technical and easiest for students to use.

Title: Birth Defects

URL: http://kidshealth.org/parent/system/ill/birth_defects.html

Description: This website provides basic information about birth defects.

Title: Basic Facts about Birth Defects

URL: www.cdc.gov/ncbddd/bd/facts.htm

Description: This website provides brief definitions, descriptions, and causes of common birth defects.

Step 1: Introduction

Duration: 30 minutes

Start the first day of this lesson by giving the pretest; then have students brainstorm what they already know about health conditions. Make a list on an overhead. This can also be done as a “free write” or a “do now.”

Present students with copies of the “Letter from the Center for Disease Defense and Prevention” or read the letter together. Go over any key vocabulary words that students may not fully understand (symptoms, birth defect, epidemiology). Explain to them that while this is a fictitious scenario, it is based on real health-related conditions that could affect many babies. Ask the students to brainstorm about what the class needs to do to take on this challenge. Write the list on the overhead. Hopefully, students’ responses should include the need for learning about common birth-related health conditions, gathering more information about the babies in these cases, checking for other patients, and working with local hospitals. If the class does not mention these topics, hint at them or make suggestions.

Supplemental Documents

Title: Pretest

File Name: Pretest.doc

Description: This Word document is used to assess students’ knowledge of key concepts presented during the lesson.

Title: Pretest Answers

File Name: Pretest Answers.doc

Description: This Word document provides the answers to the pretest.

Title: Letter from CDD

File Name: Letter from CDD.doc

Description: This is the fictitious letter sent to the school that initiates the investigation.

Step 2

Duration: 30 minutes

Prioritize the list students have developed and explain that the class starts today by researching on the Internet some of the most common birth defects.

Assign students to groups of three to five. Each group should learn about a different birth defect and report the information back to the class. Tell the students that by working in groups, the class can quickly cover a lot of information in a short period of time. The size of the group depends on your preference, the number of students in the class, and the number of computers available. One way to organize the group is to assign different jobs to the students. For a group of four students, the following jobs can be assigned:

1. Leader: Keeps the group on task and asks the teacher questions when there is a concern or problem.
2. Computer Tech: Responsible for typing in websites and searching for information.
3. Recorder: Writes down the information for the group.
4. Reporter: Reports the group's results to the rest of the class.

For additional information about cooperative groups, see the web resources below.

Assign each group one of the eight birth defects from the "Birth Defects and Developmental Disabilities Chart." The most frequent birth defects are at the top of the list. If there are fewer than eight groups, use the more frequent birth defects/developmental disabilities first.

Discuss the important information each group needs to find out about their assigned birth defect/developmental disability. Then pass out the "Birth Defects and Developmental Disabilities Chart." Clarify each of the items on the list with the class. Point out that not all birth defects/developmental disabilities have known causes, treatments, or preventive measures. The chart covers the following areas for each birth defect and developmental disability: frequency, signs and symptoms, causes, diagnosis, treatments, and prevention. Use the first example of a common cold, provided in the chart, to explain what you expect each group to do.

Remind students that the entire class is depending on each group to find the key information about a certain birth defect/developmental disability. Give the students 20–30 minutes to find the information and record their group answers. If students are experienced with Internet search engines, they can work on their own. However, if students are not familiar with Internet search engines, you can offer the web resources below as starting points for the research. During the research, monitor and observe the groups in case they become stuck on difficult vocabulary or have a hard time searching for the information. Be sure to give a count down of how much time is left for them to complete the job.

Web Resources

Title: What are some strategies for helping students work in groups?

URL: www.ncrel.org/sdrs/areas/issues/content/cntareas/science/eric/eric-7.htm

Description: This website includes strategies and key questions to ask when using group work in the classroom.

Title: Cooperative Learning

URL: www.sedl.org/scimath/compass/v01n02/1.html

Description: This website provides more information about cooperative groups in a science classroom.

Title: March of Dimes—Birth Defects

URL: www.marchofdimes.com/pnhec/4439.asp

Description: This is an organization focused on birth defects prevention. The descriptions of birth defects, developmental disabilities, and other disorders on this website are the least technical and easiest for students to use.

Title: National Center on Birth Defects and Developmental Disabilities

URL: www.cdc.gov/ncbddd/

Description: This is another website that describes most of the birth defects and developmental disabilities on the list.

Supplemental Documents

Title: Birth Defects and Developmental Disabilities Chart

File Name: Birth Defects and Developmental Disabilities Chart.doc

Description: This worksheet helps students organize the information they gather during their research.

Title: Birth Defects and Developmental Disabilities Chart Answers

File Name: Birth Defects and Developmental Disabilities Chart Answers.doc

Description: This document has the answers for the "Birth Defects and Developmental Disabilities Chart" and can be used as a reference for teachers.

Step 3

Duration: 1 hour

After the students complete their search and compile information about a given birth defect/developmental disability, have each group spend 3 minutes to present their birth defect or disability to the rest of the class. Set clear expectations for the audience and make sure to focus the time on sharing needed information. If there is time, allow for questions after each presentation.

After each group's presentation, use the overhead version of the "Birth Defects and Developmental Disabilities Chart" to discuss the answers for that particular group. Solicit answers from the group responsible for that particular birth defect/developmental disability. While students are expected to listen and record their answers during the presentations, reviewing the answers will allow them to check their answers and make sure the information needed for the last step is covered.

Conclusion

Duration: 1 hour

Now that the students have a better understanding of various birth defects and developmental disabilities, they can use this knowledge and try to diagnose some of these birth defects and developmental disabilities. Give each group a different case with a description of a baby born with a birth defect or a developmental disability. Students should use their notes to diagnose and write a report to the parents of the baby explaining their reasoning and suggestions for treatment. After investigating the case and writing their report, students should present their findings to the class. The "Case Presentation Rubric" can be used to describe the presentation format to students and assess the student presentations.

Supplemental Documents

Title: Case Files

File Name: Case Files.doc

Description: This document is a collection of birth defect cases, which is used for the final assessment.

Title: Case Files Answers

File Name: Case Files Answers.doc

Description: This document provides answers to the cases and can be used by the teacher.

Title: Medical Case Report

File Name: Medical Case Report.doc

Description: This document contains a chart that students use to report their case, which is used for the final assessment.

Title: Case Presentation Rubric

File Name: Case Presentation Rubric.doc

Description: This document contains the rubric used to assess the case study presentations.

Assessment

Students research birth defects and use their knowledge to “diagnose” birth defects in a mock investigation. Assessment takes place in the final investigation of cases, where students will be applying what they have learned about birth defects. The accuracy and justification of their responses will determine how well they have learned the material.

Modifications

Extension

Instead of doing group presentations, students can present their cases as posters or in a written form.

Students can also study more than one case if time allows.

Other Modifications

If students have not yet covered the human body, cells, and genetics, clarify any vocabulary words that may be difficult for students.

Education Standards

National Science Education Standards

As a result of their activities in grades 5–8, all students should develop understanding of SCIENCE AS INQUIRY, CONTENT STANDARD A:

- **Abilities necessary to do scientific inquiry**
- Understandings about scientific inquiry

LIFE SCIENCE, CONTENT STANDARD C:

- **Structure and function in living systems**
- **Reproduction and heredity**
- **Regulation and behavior**
- Populations and ecosystems
- Diversity and adaptations of organisms

SCIENCE IN PERSONAL AND SOCIAL PERSPECTIVES, CONTENT STANDARD F:

- **Personal health**
- **Populations, resources, and environments**
- **Natural hazards**
- **Risks and benefits**
- **Science and technology in society**

Pretest

Investigating Birth Defects Caleb Cheung, CDC's 2005 Science Ambassador Program

For each statement, write whether you think it is true or false.

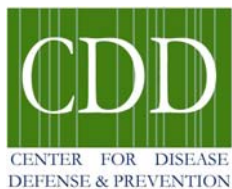
1. Epidemiology means the study of babies.
2. Babies are always born healthy and only develop problems with their bodies after they are born.
3. Babies that are born with a birth defect or disorder always have parents who have health issues.
4. Birth defects and disorders can be passed from a mom to her baby before the baby is born.
5. The most common category of birth defects is congenital heart defects.
6. A birth defect or developmental disability could be caused by the environment around the home of the family.

Pretest Answers

Investigating Birth Defects
Caleb Cheung, CDC's 2005 Science Ambassador Program

For each statement, write whether you think it is true or false.

1. Epidemiology means the study of babies.
False
2. Babies are always born healthy and only develop problems with their bodies after they are born.
False
3. Babies that are born with a birth defect or disorder always have parents who have health issues.
False
4. Birth defects and disorders can be passed from a mom to her baby before the baby is born.
True
5. The most common category of birth defects is congenital heart defects.
True
6. A birth defect or developmental disability could be caused by the environment around the home of the family.
True



Department of Health

1000 Fitness Drive, Your City, Your State 90000

October 10, 2005

7th Grade Science Students
Wellbeing Middle School
2222 22nd Ave
YourCity, CA 90000

Dear Young Scientists,

We have been informed about your work on the human body in your science classroom and would like your help in an active investigation. We recently received reports of newborn babies in your area with birth defects. So far there have been five cases of babies with various problems in the past few months. Each one of the babies has a different symptom, including a deformed lip, bluish skin, difficulty breathing, and abnormalities on the baby's back.

We have very little information about the situation and need you to investigate further. We hope that you will take on this challenging project. Please contact us if you need any additional information. Your science teacher has my contact information if you have any questions.

Sincerely,

Dr. Iwana B. Well

Dr. Iwana B. Well

Epidemiology Department
Center for Disease Defense and Prevention

Birth Defects and Developmental Disabilities Chart

Investigating Birth Defects
Caleb Cheung, CDC's 2005 Science Ambassador Program

Use the websites suggested by your teacher to fill out the information for the birth defect assigned to your group. Complete the rest of the chart during the group presentations. Certain birth defects may not have information for every column; note "none" for any column that does not have information.

1. Birth Defect or Disability Name – What is the name of the condition?
2. Frequency – How often this happens or how common is this condition? (Ex. 1 in 1,000 people)
3. Symptoms – What are the signs and symptoms of this condition?
4. Causes – What causes this condition to happen? (If known)
5. Prenatal Diagnosis – Can you know if a baby has this condition before he or she is born?
6. Treatment – What can doctors do to take care of this condition? (If available)
7. Prevention – How can this condition be prevented? (If known)

1. Name	2. Frequency	3. Symptoms	4. Causes	5. Prenatal Diagnosis	6. Treatment	7. Prevention
Example: Common cold¹	Approximately 1 in 1 adults and children	Sneezing, runny nose, sore throat, cough, possible headache, fever, and chills	Virus infection in the nose and could involve the sinuses, ears, and bronchial tubes	None	Medicine to reduce the symptoms, rest, drink water; no real cure other than letting the body fight it off	Avoid infected people, wash hands, don't touch eyes and nose with hands that might have been exposed to the virus
Congenital heart defects						

1. Name	2. Frequency	3. Symptoms	4. Causes	5. Prenatal Diagnosis	6. Treatment	7. Prevention
Autism						
Congenital hearing loss						
Sickle cell disease						
Down syndrome						

1. Name	2. Frequency	3. Symptoms	4. Causes	5. Prenatal Diagnosis	6. Treatment	7. Prevention
Cleft lip and cleft palate						
Spina bifida						
Marfan syndrome						

Reference:

1. Gwaltney J. Understanding Colds [online]. 2004. [cited 2005 Jul 20]. Available from URL: <http://www.commoncold.org/undrstn2.htm>.

Birth Defects and Developmental Disabilities Chart Answers

Investigating Birth Defects
Caleb Cheung, CDC's 2005 Science Ambassador Program

These are some of the possible answers for the "Birth Defects and Developmental Disabilities Chart." Answers may vary depending on what each group decides to present.

8. Birth Defect or Disability Name – What is the name of the condition?
9. Frequency – How often this happens or how common is this condition? (Ex. 1 in 1000 people)
10. Symptoms – What are the signs and symptoms of this condition?
11. Causes – What causes this condition to happen? (If known)
12. Prenatal Diagnosis – Can you know if a baby has this condition before he or she is born?
13. Treatment – What can doctors do to take care of this condition? (If available)
14. Prevention – How can this condition be prevented? (If known)

1. Name	2. Frequency	3. Symptoms	4. Causes	5. Prenatal Diagnosis	6. Treatment	7. Prevention
Congenital heart defects¹	1 in 125 to 150 babies	Abnormally formed heart or blood vessels leading to unusual sounding heart beats (murmur), breathing difficulty, gray or bluish skin	Not completely known, could be genetic or environmental, sometimes related to medication or illness in the mother	Listen to the baby's heart beat and look at a sonogram (picture of the heart using sound waves)	Medication, surgery to fix the problem area	Most cannot be prevented, but a woman can reduce the risk by taking vitamins and by avoiding alcohol and unprescribed drugs
Autism¹	2 to 6 children per 1,000 (boys are 3 to 4 times as likely as girls to be affected)	Difficulty interacting with others, repetitive movements, learning disabilities	Poorly understood, could be genetic or environmental causes of unusual brain development	None, diagnosed later in life	No cure, but intensive behavior treatment helps	None

1. Name	2. Frequency	3. Symptoms	4. Causes	5. Prenatal Diagnosis	6. Treatment	7. Prevention
Congenital Hearing Loss¹	3 in 1,000 babies	Lack of reaction to loud noises, which is sometimes hard to recognize in infants	Genetic, as well as non-genetic (such as some maternal infections during a pregnancy)	Some can be diagnosed prenatally, but the majority of children are diagnosed through newborn screening program	Communication skills development, hearing aids, and cochlear implants	None for genetic causes; for some infectious causes, women can get immunized
Sickle Cell Disease¹	1 in 500+ babies (mostly in African Americans and Hispanic Americans)	Pain, infections, stroke, vision problems, and slow growth	Genetic, abnormal hemoglobin in red blood cells leading to sickle shaped cells that can get stuck in blood vessels	Amniocentesis (tissue in the womb around the baby can be checked for the genetic mutation), but the majority of children are diagnosed through newborn screening program	Medicines and hospital treatments for the symptoms and complications; blood stem cell or bone marrow transplants might provide a cure	None, it is genetically inherited from parents
Down syndrome¹	1 in 800 to 1,000 babies	Varies and includes mental retardation, characteristic facial features, heart defects, infections, problems with vision and hearing	An extra chromosome number 21	Amniocentesis (tissue and fluid in the womb around the baby can be checked for the extra chromosome)	No cure; treatment for the medical problems, and providing programs to help the learning disabilities	None known, may be linked to folic acid deficiency; risk is much higher for women over the age of 35

1. Name	2. Frequency	3. Symptoms	4. Causes	5. Prenatal Diagnosis	6. Treatment	7. Prevention
Cleft lip and cleft palate¹	Cleft lip 1 in 1,000 babies Cleft palate 1 in 2,000 babies	A separation in the lips or palate (roof of the mouth)	Not well understood, could be genetic or environmental	Using a high-resolution sonogram to examine the baby's mouth and palate	Surgery by 3 months	Not well known; taking vitamins that contain folic acid before and during pregnancy may help
Spina bifida¹	1 in 2,000 babies	Ranges from no symptoms to a lump or cyst on the spine on the back, which leads to infection and paralysis; and bladder control problems	Possible combination of genetics and environmental factors causing the neural tube in the embryo not to close	Blood test may indicate a high risk, but a high-resolution sonogram or amniocentesis is needed for diagnosis	Very mild cases require no treatment, more serious cases require many surgeries, lifelong physical therapy, and care/treatment for kidney and bladder infections	Folic acid can help reduce the risk of spina bifida by up to 70% if a mother starts taking folic acid every day before she becomes pregnant
Marfan syndrome¹	1 in 5,000 babies	Tall and slender, arms and legs may be unusually long in proportion to the torso, weak aorta of the heart, displacement of the lens of the eye	Genetic, abnormal gene on chromosome 15	Difficult, in some cases can be identified with genetic testing of a tissue sample obtained from amniocentesis	Monitoring the eyes and circulatory system, having surgery on the heart or eyes, if necessary	None

Reference:

1. March of Dimes. Birth Defects and Genetic Conditions [online]. 2005. cited 2005 July 20]. Available from URL: <http://www.marchofdimes.com/pnhec/4439.asp>

Additional resources:

Autism

Centers for Disease Control and Prevention. Autism [online]. 2005. Available from URL: <http://www.cdc.gov/ncbddd/autism/>

Congenital hearing loss

Centers for Disease Control and Prevention. Congenital hearing loss [online]. 2005. Available from URL:

<http://www.cdc.gov/ncbddd/dd/ddhi.htm>

<http://www.cdc.gov/ncbddd/ehdi/question.htm>

<http://www.cdc.gov/ncbddd/ehdi/FAQ/questiontreatment.htm#intervention>

http://www.asha.org/public/hearing/disorders/how_know.htm

Down syndrome

Centers for Disease Control and Prevention. Down syndrome [online]. 2005. Available from URL:

<http://www.cdc.gov/ncbddd/bd/ds.htm>

Spina bifida

Centers for Disease Control and Prevention. Spina bifida [online]. 2005. Available from URL:

<http://www.cdc.gov/ncbddd/folicacid/faqs.htm#spina>

Spina Bifida Association. Frequently Asked Questions About Spina Bifida [online]. 2005.

Available from URL: http://www.sbaa.org/site/PageServer?pagename=ASB_faq

Case Files
Investigating Birth Defects
Caleb Cheung, CDC's 2005 Science Ambassador Program

Teachers: Cut the following cases along the dotted lines and give one per group to use for their medical case report as a final assessment.

CASE #1

A parent brings in his 6-month-old son, Jerome. He is concerned because Jerome's weight is lower than normal and he has had several infections in the past few months. Jerome's grandparents were from Ethiopia.

CASE #2

A parent brings in her 2-year-old son, John. She is concerned because John hasn't begun speaking yet and has difficulty interacting with other children when they play together. He also has a lot of repetitive movements and avoids making eye contact with other people. He has an older brother with similar symptoms.

CASE #3

A father brings in his 6-month-old son, Ken. He is concerned because Ken doesn't seem to pay attention and doesn't respond to music like their first child. He also mentions that Ken's grandfather used a hearing aid all his life.

CASE #4

A mother calls you about her newborn baby, Sonia. The mother and her doctor are concerned because Sonia has unusual facial features and a large tongue. She is also very flexible with little muscle tone.

CASE #5

A father contacts your office about his newborn daughter, Elizabeth. Elizabeth's parents and doctors are concerned because there is something unusual about Elizabeth's upper lip. It looks as if there is a separation between the right and left sides of her lip.

CASE #6

A doctor contacts you about a newborn baby named Dewanna. She is concerned because Dewanna has a red opening on the lower part of her back. Dewanna's mother reports that she never consumed folic acid or a multivitamin during the pregnancy.

CASE #7

A family brings in their 2½-year-old son, Aaron. They are concerned because Aaron appears to have really long arms compared to his siblings. The mom also tells you that many people in their extended family are very tall. Aaron also has flat feet and has very flexible joints.

CASE #8

A father brings in his 1-week-old daughter, Nancy. He is concerned because Nancy recently started having difficulty breathing and her skin looks blue. He also tells you that Nancy's mom drank alcohol during the pregnancy.

Case Files Answers

Investigating Birth Defects
Caleb Cheung, CDC's 2005 Science Ambassador Program

Here are the answers to the case files.

Case Description	Diagnosis	Age	Gender	Possible causes	Prenatal Diagnosis	Treatments	Prevention
CASE #1 A parent brings in his 6-month-old son, Jerome. He is concerned because Jerome's weight is lower than normal and he has had several infections in the past few months. Jerome's grandparents were from Ethiopia.	Sickle cell disease	6 months	Male	Genetic, abnormal hemoglobin in red blood cells leading to sickle shaped cells that can get stuck in blood vessels	Amniocentesis (tissue in the womb around the baby can be checked for the genetic mutation), but the majority of children are diagnosed through newborn screening program	Medicines and hospital treatments for the symptoms and complications; blood stem cell or bone marrow transplants might provide a cure	None, it is genetically inherited from parents
CASE #2 A parent brings in her 2-year-old son, John. She is concerned because John hasn't begun speaking yet and has difficulty interacting with others children when they play together. He also has a lot of repetitive movements and avoids making eye contact with other people. He has an older brother with similar symptoms.	Autism	2 years old	Male	Poorly understood. There could be genetic or environmental causes of the unusual brain development seen in autism.	None, diagnosed later in life	No cure, but intensive behavior treatment helps	None
CASE #3 A father brings in his 6-month-old son, Ken. He is concerned because Ken doesn't seem to pay attention and doesn't respond to music like their first child. He also mentions that Ken's grandfather used a hearing aid all his life.	Congenital hearing loss	6 months	Male	Genetic, as well as non-genetic (such as some maternal infections during a pregnancy)	Some can be diagnosed prenatally, but the majority of children are diagnosed through newborn screening program	Communication skills development, hearing aids, and cochlear implants	None for genetic causes; for some infectious causes, women can get immunized
CASE #4 A mother calls you about her newborn baby, Sonia. The mother and her doctor are concerned because Sonia has unusual facial features and a large tongue. She is also very flexible with little muscle tone.	Down syndrome	Newborn	Female	An extra chromosome number 21	Amniocentesis	No cure; treatment for the medical problems, and providing programs to help the learning disabilities	Not known, maybe linked to folic acid deficiency; risk much higher for women over 35

CASE #5 A father contacts your office about his newborn daughter, Elizabeth. Elizabeth's parents and doctors are concerned because there is something unusual about Elizabeth's upper lip. It looks as if there is a separation between the right and left sides of her lip.	Cleft lip and cleft palate	Newborn	Female	Not well understood, could be genetic or environmental	Using a high-resolution sonogram to examine the baby's mouth and palate	Surgery by 3 months	Not well known; taking vitamins that contain folic acid before and during pregnancy may help
CASE #6 A doctor contacts you about a newborn baby named Dewanna. She is concerned because Dewanna has an red opening on the lower part of her back. Dewanna's mother reports that she never consumed folic acid or a multivitamin during pregnancy.	Spina bifida	Newborn	Female	Possible combination of genetics and environmental factors	Blood test may indicate a high risk, but a high-resolution sonogram or amniocentesis is needed for diagnosis	Very mild cases require no treatment, more serious cases require many surgeries, lifelong physical therapy, and care/treatment for kidney and bladder infections	Folic acid can help reduce the risk of spina bifida by up to 70% if a mother starts taking folic acid every day before she becomes pregnant
CASE #7 A family brings in their 2½-year-old son, Aaron. They are concerned because Aaron appears to have really long arms compared to his siblings. The mom also tells you that many people in their extended family are very tall. Aaron also has flat feet and very flexible joints.	Marfan syndrome	2.5 years old	Male	Genetic, abnormal gene on chromosome 15	Difficult, in some cases can be identified with genetic testing of a tissue sample obtained from amniocentesis	Monitoring the eyes and circulatory system, having surgery on the heart or eyes, if necessary	None
CASE #8 A father brings in his 1-week-old daughter, Nancy. He is concerned because Nancy recently started having difficulty breathing and her skin looks blue. He also tells you that Nancy's mom drank alcohol during the pregnancy.	Congenital heart defects	1 week	Female	Not completely known, could be genetic or environmental, sometimes related to medication or illness in the mother	Listen to the baby's heart beat and look at a sonogram (picture of the heart using sound waves)	Medication, surgery to fix the problem area	Most cannot be prevented, but a woman can reduce the risk by taking vitamins and by avoiding alcohol and unprescribed drugs

Reference:

March of Dimes. Birth Defects and Genetic Conditions [online]. 2005. [cited 2005 July 20]. Available from URL: <http://www.marchofdimes.com/pnhec/4439.asp>

Additional resources:

Autism

Centers for Disease Control and Prevention. Autism [online]. 2005. Available from URL: <http://www.cdc.gov/ncbddd/autism/>

Congenital hearing loss

Centers for Disease Control and Prevention. Congenital hearing loss [online]. 2005. Available from URL:

<http://www.cdc.gov/ncbddd/dd/ddhi.htm>

<http://www.cdc.gov/ncbddd/ehdi/question.htm>

<http://www.cdc.gov/ncbddd/ehdi/FAQ/questiontreatment.htm#intervention>

http://www.asha.org/public/hearing/disorders/how_know.htm

Down syndrome

Centers for Disease Control and Prevention. Down syndrome [online]. 2005. Available from URL:

<http://www.cdc.gov/ncbddd/bd/ds.htm>

Spina bifida

Centers for Disease Control and Prevention. Spina bifida [online]. 2005. Available from URL:

<http://www.cdc.gov/node.do?id=0900f3ec80010af9>

Medical Case Report

Investigating Birth Defects

Caleb Cheung, CDC's 2005 Science Ambassador Program

For this assignment, your group will pretend to be doctors. Read the medical case together as a group. Try to figure out what birth defect or developmental disability your patient might have. Using your notes, write a report that will be submitted to the CDD to help them with their ongoing investigation.

Child's Name:	Gender:	Age:
Description of Symptoms:		
Diagnosis: (Include possible birth defects and/or developmental disabilities)		
Possible Causes:		
Suggestions to the Family (tests, treatments, resources):		
Report Submitted by: (Doctors' Names)		
Signatures:		

Case Presentation Rubric

Investigating Birth Defects
Caleb Cheung, CDC's 2005 Science Ambassador Program

Now that you and your fellow doctors have discussed your assigned case and made conclusions, it is time to share your case with the class. For your presentation, your group should:

- Briefly summarize your case
- Discuss how you decided on your diagnosis
- Give a short (1–2 minute) summary of the condition that you diagnosed, including possible causes
- Share the recommendations that you made to the family

You should be able to present your case in 5 to 10 minutes. Be sure to use the rubric below to ensure that your presentation will receive the highest number of points possible. You will also be scored on how well you pay attention to the other presentations, so be sure to be a good listener!

		2	1
Organization	Presentation was well-organized and easy to follow. Information was presented in a concise manner and the presentation contained all of the required components.	Presentation was organized and easy to follow most of the time. The presentation was missing a required component or was not concise.	Presentation was not organized and did not include one or more of the required components.
Accuracy	The case was well-researched; the patient was diagnosed correctly, and the summary of the condition and recommendations to the family were accurate.	The information presented had one or two small inaccuracies, or the patient was diagnosed incorrectly, but the group was able to justify their diagnosis.	The case was not researched well; the patient was diagnosed incorrectly and the group could not justify their diagnosis, and incorrect information was presented.
Group Collaboration	Student almost always listens to, shares with, and supports the efforts of others in the group. Tries to keep people working well together.	Often listens to, shares with, and supports the efforts of others in the group but sometimes is not a good team member.	Rarely listens to, shares with, or supports the efforts of others in the group. Often is not a good team member.
Listening skills	Listens intently. Does not make distracting noises or movements.	Sometimes does not appear to be listening but is not distracting.	Sometimes does not appear to be listening and makes distracting noises or movements.